

## **Sex Chromosomes and Sex determination**

### **Sex Chromosomes:**

Most animals show sexual dimorphism; in other words, an individual can be either male or female.

In most of these cases, sex is determined by special sex chromosomes. In these organisms, there are two categories of chromosomes, sex chromosomes and autosomes (the chromosomes other than the sex chromosomes).

Most of the chromosomes in genome are autosomes. The sex chromosomes are fewer in number and majority of genes on the sex chromosomes are also not directly involved in sex determination.

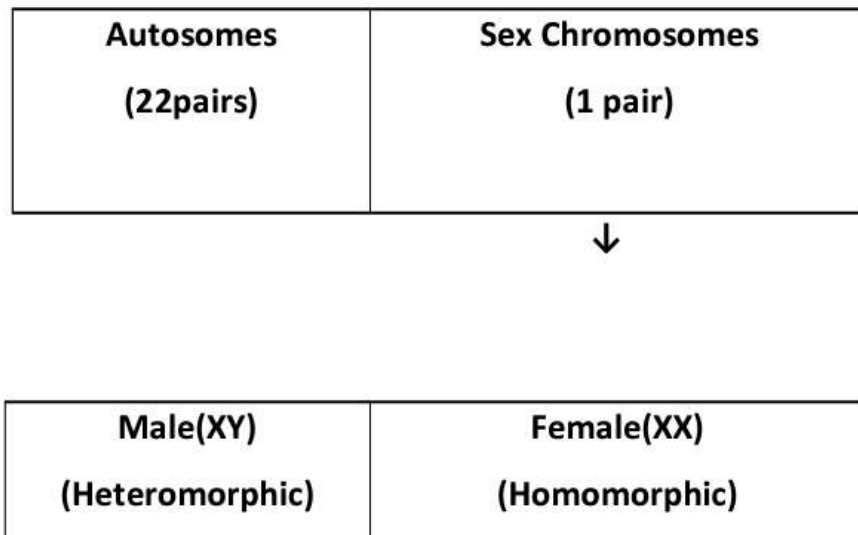
In human body cells have 46 chromosomes: 22 homologous pairs of autosomes plus 2 sex chromosomes.

There are two types of sex chromosomes in human-X and Y. In females, there is a pair of X-chromosomes. In males, there is a non-identical pair, consisting of one X and one Y.

Hence, human females are homomorphic and males are heteromorphic. The Y- chromosome is considerably shorter than the X. The X and Y – chromosomes Harbor different numbers of genes.

The Human Genome Project has identified 397 possible genes on the human Y-chromosome, but fewer than 100 of them seem to be functional. By comparison, it has identified more than 1000 genes on the human X-chromosome.

**Human nuclear chromosomes:**



During meiosis in females, the two X-chromosomes pair and segregate like autosomes so that each female gamete receives one X-chromosome, hence, the female is said to be the homogametic sex.

During meiosis in males, the X and Y- chromosome pair and segregate so that half of the male gametes receive X and the other half receive Y. Therefore, the male is said to be the heterogametic sex.

The human Y- chromosome can be divided structurally into three regions:

1. Male –specific region of the Y- chromosome,
2. Pseudoautosomal regions (PAR1 and PAR2),and
3. Heterochromatin region.

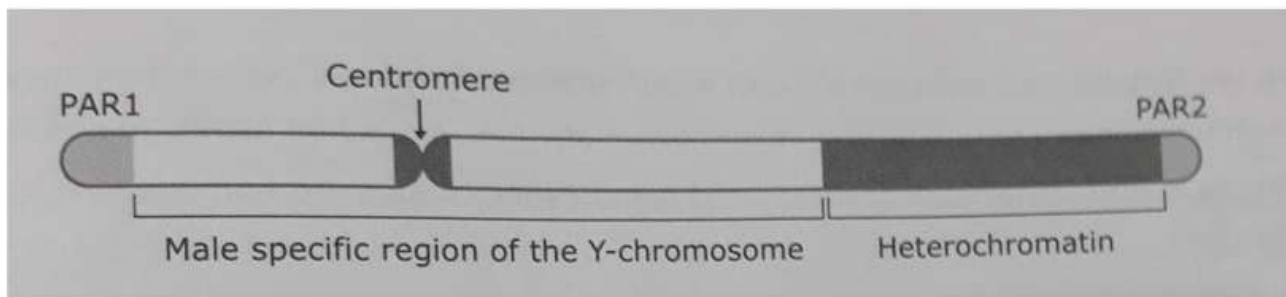


Fig. Schematic diagram of human Y-chromosome indicating the protein-coding genes.

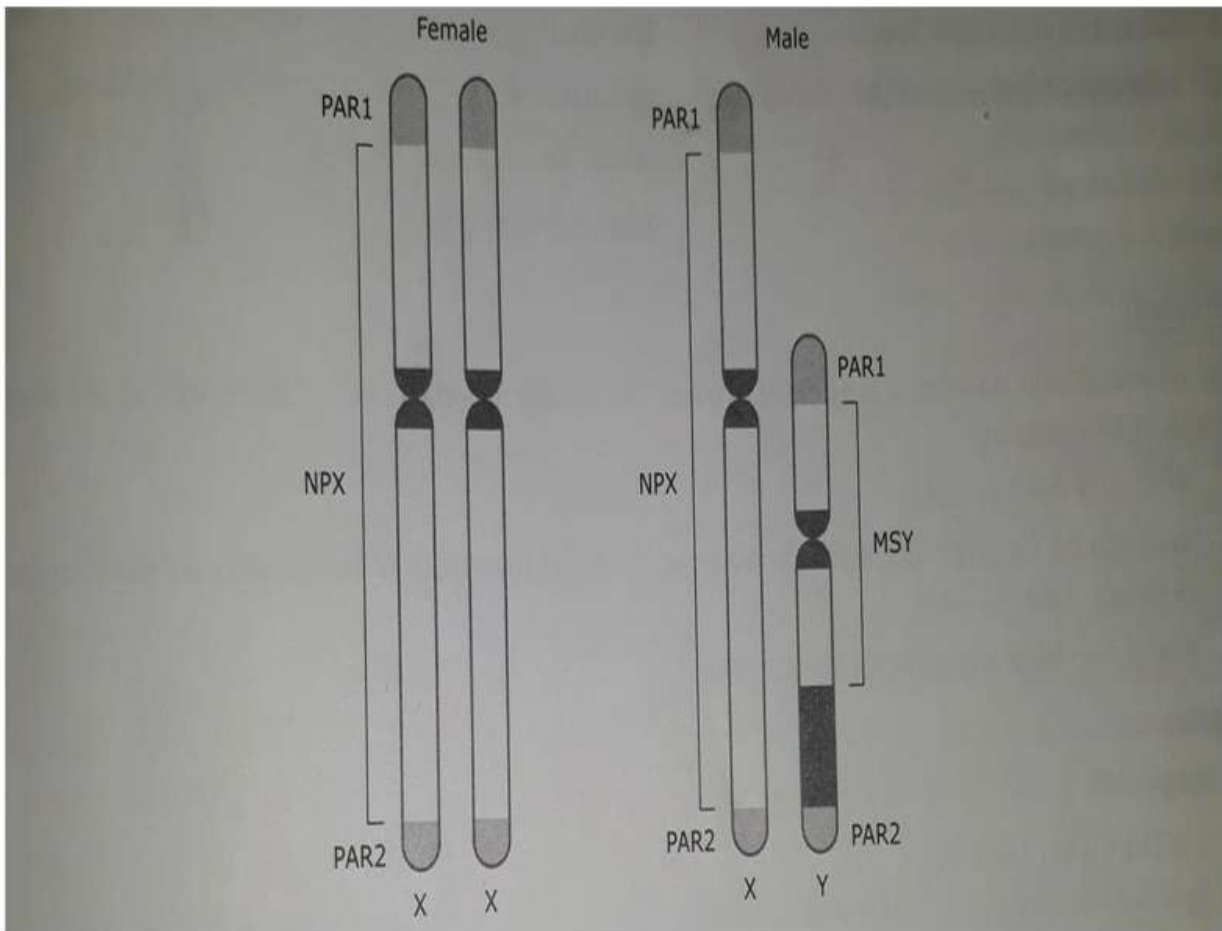
PARs contain 20 protein- coding genes (16 genes in PaR1 and 4genes in PAR2).PARs are present in both X and Y chromosomes.

The pairing of the X and Y –chromosomes is made possible by a major pseudoautosomal region (PAR1) of 2.6 Mb located at the tips of the short arms of both chromosomes and a minor pseudoautosomal region (PAR2) of 320kb locked at the tips of the long arms of both chromosomes.

Genes in the PAR1 segment have some interesting properties: they are not subject to X-inactivation and because of the crossing over, alleles at these loci do

not show the normal X-linked or Y-linked patterns of inheritance, but segregate like autosomal alleles.

The male- specific region of the Y-chromosome contains 23 protein-coding genes and numerous pseudogenes.



**Fig. At each end of the human X and Y- chromosomes are the PARs, which recombine during meiosis and therefore contain the same genes.**

### Sex determination in animal:

Whether an animal will become a male, a female, or a hermaphrodite is determined very early in development.

Hermaphrodites are individuals that contain both male and female sex organs. Hermaphroditism is rare among animals.

Some animals such as gastropods and earthworms are simultaneous hermaphrodites whereas many fishes are sequential hermaphrodites.

Sex- determining mechanisms in animals are diverse. These mechanisms may be genotypic and environmental.

### **Genotypic sex determination:**

In genotypic sex determination, an individual's sex established by its genotype (e.g. mammals, birds, amphibians, most insects, some reptiles and fish).

In many animals, presence or absence of a particular chromosome or number of chromosomes determines the male and female sex.

Basically, four types of chromosomal sex – determining mechanisms exist in animals; the XY, ZW, XO and haplodiploid system.

i. **XY system:**

In the XY system, the females are homogametic with a pair of X-chromosomes (XX) and males are heterogametic with one X and Y-chromosomes (XY).

ii. **ZW system:**

Almost all mammals, many files and some fishes, males are heterogametic. In the ZW system, as in birds, snakes, butterflies, and



some fishes; males are homogametic (ZZ) , and females are heterogametic (ZW).

**iii. XO system:**

In the XO system, as in grasshopper; males have only one X-chromosome (XO) and females have two X-chromosomes (XX).

**iv. Haplodiploid systems:**

In haplodiploid systems, as in honey bees, male progeny normally develops from unfertilized eggs, which are haploid and have just one set chromosomes.

The fertilized honey bee eggs, which are diploid and have two sets of chromosomes, differentiate into queens and worker bees.

Thus , in honey bees, sex is determined by the fertilization or non-fertilization of eggs, rather than the presence or absence of sex chromosomes.

**Different systems of sex determination:**

1. XY system: (e.g. Human) XX(female) XY (male)
2. XO system :(e.g. Grasshopper) XX (female) XO (male)
3. ZW system :( e.g. Birds) ZW (female) ZZ(male)
4. Haplodiploid system:(e.g. Honey bees) Diploid(female) Haploid (male, drones) Worker bee (imperfect female) Queen(perfect female)

**Environmental sex determination:**

Sex is also determined by environmental factors, such as temperature, pH, social interactions and seasonality.

In many species, sex is determined by the temperature at which the egg is incubated during a temperature sensitive period and cannot be predicted by zygotic genotype. This peculiar mechanism is termed temperature – dependent sex determination.

The temperature dependent sex determination has been shown to exist in all crocodiles and the majority of turtles along with a few species of lizards.

In these reptiles, the temperature of eggs during a certain period of development is the deciding factor in determining sex, and small changes in temperature can cause dramatic changes in the sex ratio.

Often, eggs incubated at low temperatures produce one sex, whereas eggs incubated at higher temperatures produce the other.

### **Sex determination in mammals:**

The sex of eutherian (or placental) mammal is determined by its sex chromosomes. Primary sex determination is dictated by whether an organism has an XX or XY karyotype.

Female have two X-chromosomes in all of their somatic cells whereas males have one X and one Y. individuals with a Y – chromosome develop as males no matter how many X- chromosomes they have , whereas individuals without a Y- chromosome develop as females.

Thus, primary sex determination is based on the presence of the Y- chromosome. Primary sex determination is the determination of the gonads – the egg-forming ovaries or sperm- forming testes.

In contrast, secondary sex determination is the determination of the male or female phenotype by the hormones produced by the gonads.

### **Genic balance theory of sex determination in Drosophila:**

Fruit flies also have XX females and XY males. However, the mechanism of sex determination in Drosophila differs from that in mammals.

In mammals, the Y- chromosome plays a pivotal role in determining the male sex. In Drosophila, the Y- chromosome is not involved in determining sex.

Rather, in Drosophila, genes present on the Y-chromosome involved in sperm formation in adults, but not in sex determination.

Calvin Bridges suggested in 1921 that sex in Drosophila is determined by the balance (ratio) of autosomal alleles that favour maleness and alleles on the X- chromosomes that favour femaleness.

He found that a ratio of X- chromosomes to autosomal sets determines the sex of Drosophila. A normal diploid male contains 2 sets of autosomes and XY- chromosomes. Similarly, a normal diploid female has 2 sets of autosomes and two X- chromosomes.



### Sex determination by genic balance in Drosophila:

Number of X-chromosome	Number of Autosomal sets (A)	Total number of Chromosomes	X/A ratio	Sex
3	2	9	1.5	Meta female
2	2	8	1.00	Female
1	1	4	1	Female
2	3	11	0.67	Intersex
1	2	7	0.50	Male
1	3	10	0.33	Meta male

Bridges results show that in Drosophila factors that cause a fly to develop into a male are not localized on the sex chromosome, but are instead found on the autosomes.

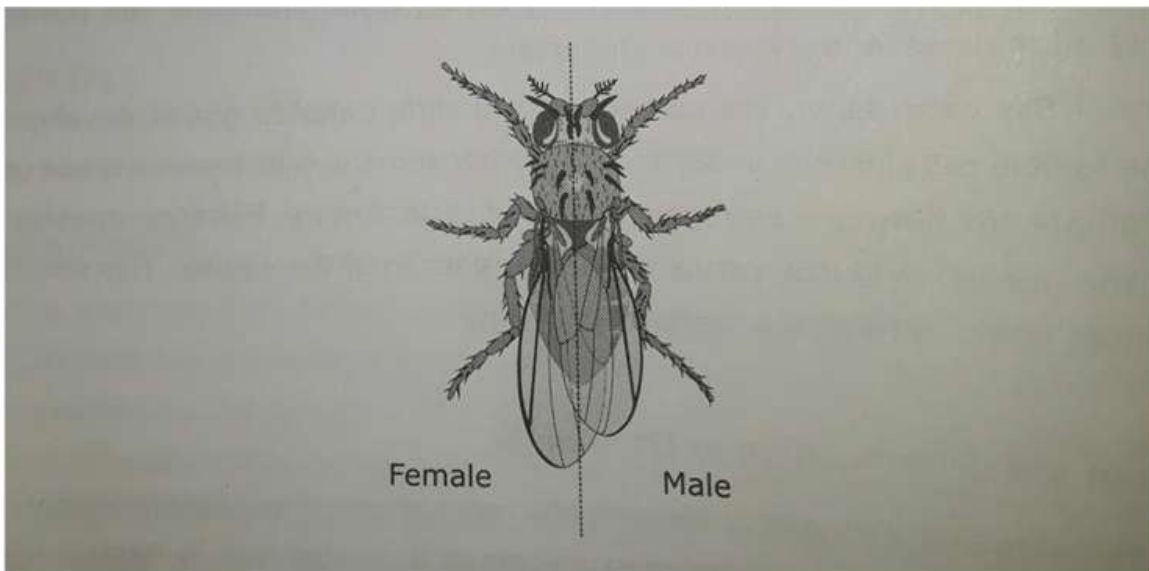
X-chromosomes contain genes for some female-determining factors. A sex switch gene directs the female development. This sex switch gene termed sex-lethal (Sxl) is located on the X-chromosome.

In the on state, it diverts female development and in the off state maleness. Other genes located on the X- chromosome and the autosomes regulate this sex switch gene. Activation of the Sxl gene relies on a ratio of X- chromosomes to sets of autosomes.

The presence of the Y- chromosome in *Drosophila*, although it is essential for male fertility, has nothing to do with the determination of sex.

### ***Drosophila* gynandromorphy:**

A gynandromorphy is an organism that contains both male and female characteristics. A gynandromorph can have bilateral asymmetry, half female and half male, or they can be mosaic. A classic example is the *Drosophila* gynandromorphy.



**Fig. Gynandromorphs in *Drosophila*.**